学位論文の要旨

# Fetal outcome of trisomy 18 diagnosed after 22 weeks of gestation: Experience of 123 cases at a single perinatal center

# 妊娠 22 週以降に診断された 18 トリソミー症例の予後について

## ―123 症例の臨床解析の結果から―

Hiromi Nagase

長瀬　寛美

Obstetrics and Gynecology

Yokohama City University Graduate School of Medicine

横浜市立大学大学院医学研究科

生殖生育病態医学（産婦人科）

Doctoral Supervisor: Fumiki Hirahara Professor

指導教員：平原史樹 教授

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## 【Introduction】

Trisomy 18 is an autosomal chromosomal abnormality syndrome caused by the duplication of all or part of chromosome 18, which was reported first in Lancet (Edwards et. al. 1960). The mortality rate has been reported as approximately 56 ~61 % within a month after birth and 90~95% of all trisomy 18 infants have been reported to die within one year of birth. (Imataka et al., 2007; Rasmussen et al., 2003) And the median survival time has been reported 3-15 days. (Embleton et al., 1996; Rasmussen et al., 2003)　Because of the morality rate, withholding or withdrawal of intensive treatment had been considered and the palliative care had been recommended to the parents. (Carter et al., 1985; Bos et al., 1992; Embleton et al., 1996; Jones, 2006). Recently, some reports have been published about the intensive care and the surgical procedures for the infants with trisomy 18, concluding that intensive care and surgical procedures could improve the prognosis of infants with trisomy 18 (Graham et al. ,2004; Kosho et al., 2006,2013; Kaneko et al., 2008, 2009; Iwami et al., 2011; Maeda et al., 2011; Nishi et al., 2013). To discuss about the management of pregnancy with a

fetus affected with trisomy 18, we have to inform the parents the prognosis of the fetus/infant when they choose C-section, when they choose vaginal delivery, when they choose palliative care for the fetus/infant or intensive care for the infant. The parents should decide autonomously by the appropriate and the accurate medical information about the trisomy 18.

Here, we described, from a series of 123 fetuses/infants born and treated in our hospital, clinical features and survival of them managed basically through modern medical care and palliative approach both for the mothers and fetuses/infants.

【Material and Methods】

We studied 123 patients with trisomy 18 who were born after 22 weeks of gestation at the Kanagawa Children's Medical Center (a tertiary care referral center) from 1993 to 2009 using a retrospective chart review. The statistical analysis was performed by chi-square test or t-test using SPSS$^{R}$ ver. 21. All of the cases were diagnosed as trisomy 18 by a chromosomal analysis. All of the patients underwent a detailed ultrasound scan by both obstetricians and neonatologists. The diagnosis of trisomy 18 was confirmed by antenatal or postnatal chromosomal analysis. When we encountered a fetus who was diagnosed or strongly suspected as trisomy 18, the obstetricians, neonatologists, medical geneticists, midwives, nurses and medical social workers discussed the medical findings and how to support the parents when making decisions. Then we informed the parents of the natural history of trisomy 18. During this studying period, we recommended to avoid

invasive procedures for the mother or infant based on fetal indications.

【Results】

Among the 123 cases, 95.9% were diagnosed with trisomy 18 prenatally. Prenatal ultrasound findings showed fetal growth restriction in 77.2%, polyhydramnios in 63.4% and congenital heart defects in 95.1%. For 18 cases, Caesarean section (C-section) was chosen, and for 75 cases, transvaginal delivery was chosen. Premature delivery occurred in 35.5%. Stillbirths occurred in 50 cases (40.7%). Fetal demise before onset of labor occurred in 30 cases and fetal demise during labor occurred in 20 cases which was 26.7% of vaginal deliveries. The median survival time of live-born infants was 3.5 days. Early neonatal (<7 days of age ) death occurred in 39 cases (53% of live-born infants), late neonatal death (7-27 days of age ) occurred in 8 cases (11% of live-born infants), infantile (28-364 days of age) death occurred in 21 cases (29% of live-born infants), and 2 cases (3% of live-born infants) were considered to be “long-term survivors” following survival for more than 3 years Among the live-born infants, the survival rate for 24 hours, 1 week, 1 month and 1 year were 63%, 43%, 33% and 3%. The median survival time was 3.5 days. There was no significant difference between the survival time of C-section and that of vaginal delivery. However, for the births involving breech presentation, the survival time of C-section was significantly longer than that of vaginal delivery.

【Discussion】

The prognosis of infants with trisomy 18 is not optimistic and is influenced by many factors such as associated anomalies, gestational week, birth weight ,mode of delivery, or even by the available medical resources. Therefore the management for the cases of trisomy 18 should be individualized. When the parents choose palliative care for the infants, we put special emphasis on the baby's time to spend with the family.

【Conclusion】

These data are used in prenatal counseling to the couples whose fetuses have been found to have trisomy 18, as the prognosis of the fetuses/infants of trisomy 18 at the hospital where palliative care tends to be chosen and invasive procedures tend to be avoided.

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論文目録

Ⅰ　主論文

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